Letter to the Editor

Response to Drs. Shastry and Trese: Phenotype-Genotype Correlations in X-Linked Retinitis Pigmentosa

To the Editor:

Shastry and Trese [1995] recently reported on a large kindred with X-linked retinitis pigmentosa (XLRP) characterized by a loss of central vision and preserved peripheral function. In their report, the disease had an early onset with severe myopia and a loss of central vision, while night blindness occurred later. Genetic analysis suggested that the disease was linked to the RP2 locus, and the authors raised the question of whether other cases linked to RP2 could display a similar loss of central vision. Three years ago, we reported on 4 large XLRP pedigrees with a very early onset with severe myopia and early loss of visual acuity, while in 5 other families the disease started later with night blindness. We showed that the first clinical form was linked to RP2, while the second was linked to RP3 [Kaplan et al., 1992]. Thus, the major difference between the two forms concerns the initial symptom, information which can be obtained from the parents and patients after careful questioning. By contrast, in adult life, no difference in either severity of disease or aspect of the fundus was observed in our series, regardless of the clinical subtype of XLRP. Some months later, Jacobson et al. [1992] reported on a pedigree with an RP2 genotype, and their data support the notion that in XLRP of RP2 type 1, cone dysfunction takes place first, and as the disease advances both rods and cones are affected. We were very happy, therefore, to read that the study of Shastry and Trese [1995] fully confirmed our previous findings.

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Received for publication December 30, 1995; revision received January 26, 1996.

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